DNAH5 gene

dynein axonemal heavy chain 5

Normal Function

The *DNAH5* gene provides instructions for making a protein that is part of a group (complex) of proteins called dynein. This complex functions within cell structures called cilia. Cilia are microscopic, finger-like projections that stick out from the surface of cells. Coordinated back and forth movement of cilia can move the cell or the fluid surrounding the cell. Dynein produces the force needed for cilia to move.

Within the core of cilia (the axoneme), dynein complexes are part of structures known as inner dynein arms (IDAs) or outer dynein arms (ODAs) depending on their location. Coordinated movement of the dynein arms causes the entire axoneme to bend back and forth. IDAs and ODAs have different combinations of protein components (subunits) that are classified by weight as heavy, intermediate, or light chains. The *DNAH5* gene provides instructions for making heavy chain 5, which is found in ODAs. Other subunits are produced from different genes.

Health Conditions Related to Genetic Changes

Primary ciliary dyskinesia

More than 80 mutations in the *DNAH5* gene have been found to cause primary ciliary dyskinesia, which is a condition characterized by respiratory tract infections, abnormal organ placement, and an inability to have children (infertility). *DNAH5* gene mutations result in an absent or abnormal heavy chain 5. Without a normal version of this subunit, the ODAs cannot form properly and may be shortened or absent. As a result, cilia cannot produce the force needed to bend back and forth. Defective cilia lead to the features of primary ciliary dyskinesia.

Heterotaxy syndrome

MedlinePlus Genetics provides information about Heterotaxy syndrome

Other Names for This Gene

- axonemal beta dynein heavy chain 5
- CILD3

- ciliary dynein heavy chain 5
- DNAHC5
- DYH5_HUMAN
- dynein heavy chain 5, axonemal
- dynein, axonemal, heavy chain 5
- dynein, axonemal, heavy polypeptide 5
- FLJ46759
- HL1
- KIAA1603

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DNAH5 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1767[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28DNAH5%5BTIAB%5D%29+OR+%28DNAHC5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- DYNEIN, AXONEMAL, HEAVY CHAIN 5; DNAH5 (https://omim.org/entry/603335)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1767)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DNAH5[gene])

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Genomic Location

The *DNAH5* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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